Research Brief

A SUMMARY OF A PUBLISHED ARTICLE

Trajectories and Predictors of the Development of Very Young Boys with FXS

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FINDINGS FROM FAMILY ADAPTATION TO FXS, A STUDY CONDUCTED AT THE UNIVERSITY OF NORTH CAROLINA AT CHAPEL HILL

Fragile X syndrome (FXS), the most common inherited cause of developmental disabilities, can be diagnosed prenatally or at birth with genetic testing; however, the average age of diagnosis is 32 months when there is no known family history. As a result, many young children with FXS do not receive early treatment to address developmental delays. One barrier to early detection of FXS is the lack of information regarding the characteristics of FXS in infants and toddlers.

Since most children with FXS are not diagnosed until they are nearly 3 years old, research on young children with FXS is limited. However, a few studies have looked at infants 12 months of age or younger and suggest that developmental delays (i.e., language delays) or specific behaviors (i.e., sensory or regulatory) may be detectable in infants with FXS within the first year. As children with FXS age, these developmental delays or atypical behaviors often become more pronounced. Although these preliminary findings contribute to the literature and provide a foundation for further research, these studies have included small samples, focused on narrow age ranges, or used limited assessment measures. Additional research is essential to understanding the development of infants with FXS.

The primary goal of this study was to describe the trajectories and examine predictors of development for boys with FXS during the first 5 years of life. This study also attempted to identify the age at which development was clearly delayed and the extent to which early developmental scores were associated with later developmental scores.

Participants for this study included 55 boys, aged 8 to 68 months, with FXS. All boys were assessed at least twice and most were assessed 3 or more times for a total of 189 assessments across the participants. The Mullen Scales of Early Learning (MSEL) was used to assess development.

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- Participants demonstrated stead developmental gains across the first five years of life with no evidence of a decline in the rate of development during this age period.
- All 4 domains of development (fine motor, receptive language, expressive language, visual reception) appeared delayed; however, fine motor appeared most severely delayed at this time and expressive language was more delayed than receptive language.
- The degree of autistic behavior was a strong predictor of developmental outcomes, particularly to language outcomes.
- Delays first emerged at 9 months of age.
- Early developmental skills were moderately related to later scores with age and autistic behavior the most powerful determinants.

Overall, boys with FXS between the ages of 8 to 68 months displayed developmental delays in all five categories of the MSEL. The rate of development, however, appeared to remain stable with no decline during this age period. Fine motor skills were most delayed in this sample of young boys, and expressive language was more delayed than receptive language. Developmental delays were evident as early as 9 months in language categories of the MSEL. Children with less autistic behavior displayed a faster rate of development over time. Although most children in this sample who exhibited autistic behavior did not meet diagnostic criteria for autism, the presence autistic-like behaviors did relate to several areas of development, particularly language skills.

Findings from this study provide vital information about the developmental characteristics of infants with FXS. These characteristics may also serve as identification markers for infants who have yet to be diagnosed. Based on the results of this study, the following recommendations should be taken into consideration in order to improve detection of FXS and overall development of children with FXS. First, medical practitioners should refer infants diagnosed with FXS to early intervention services, even if developmental delays are not apparent, in order to minimize delays that may emerge in the early years. Second, routine developmental screening for all young children should be implemented to detect delays in infants with FXS and other conditions. Third, children with FXS should be tested for autism due to its high rate of co-occurrence with FXS and the more severe developmental outcomes that may accompany autistic symptoms. If autistic symptoms are present, treatments known to be effective for children with autism may also benefit children with co-morbid autism and FXS.

This research summary is based on the following published article: Roberts JE, Mankowski J, Hatton DD, Baranek GT, Mirrett PL, Reznick JS, Long AC, Bailey DB. Trajectories and predictors of the development of very young boys with fragile X syndrome. Journal of Pediatric Psychology. 2009; 4: 827-36.This summary was prepared by the Fragile X Research Registry. If you have any questions or would like to contact the researchers of this study, please send an email to info@FragileXRegistry.org.